Genetics Uncoded:

Facts about

HFE-Associated Hereditary Hemochromatosis

What Your Test Results Mean

Carriers typically show no symptoms of *HFE*-associated hereditary hemochromatosis (HFE-HH); however, carriers are at an increased risk of having a child with HFE-HH. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

HFE-HH Explained

HFE-HH is an inherited condition that causes the body to absorb too much iron from the diet. The excess iron is stored in the body's tissues and organs, particularly the skin, heart, liver, pancreas, and joints. Excess iron cannot be excreted properly, thus causing damage to tissues and organs. Early symptoms may include fatigue, joint pain, abdominal pain, and loss of sex drive. Later signs and symptoms may include arthritis, liver disease, diabetes, heart abnormalities, and skin discoloration. In rare cases, symptoms may begin before birth, resulting in liver damage that is apparent at birth or within the first few days of life. Treatment includes therapeutic phlebotomy, iron chelation therapy, and dietary changes.

How the Genetics Work

HFE-HH is an autosomal recessive disorder caused by pathogenic variants of the *HFE* gene. In general, individuals have two copies of the *HFE* gene. Carriers have a single variant in one copy of the HFE gene while individuals with HFE-HH have variants in both copies of *HFE*, one inherited from each parent. Risk for two carriers to have a child with HFE-HH is 25%.

Questions? Contact us at **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.

Clarity Genetics, LLC
5231 NW 33rd Ave
Ft. Lauderdale, FL 33309
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